FREM1 gene

FRAS1 related extracellular matrix 1

Normal Function

The *FREM1* gene provides instructions for making a protein that is involved in the formation and organization of basement membranes, which are thin, sheet-like structures that separate and support cells in many tissues.

The FREM1 protein is one of a group of proteins, including proteins called FRAS1 and FREM2, that interact during embryonic development as components of basement membranes. Basement membranes help anchor layers of cells lining the surfaces and cavities of the body (epithelial cells) to other embryonic tissues, including those that give rise to connective tissues (such as skin and cartilage) and the kidneys.

Health Conditions Related to Genetic Changes

Manitoba oculotrichoanal syndrome

At least two *FREM1* gene mutations have been identified in people with Manitoba oculotrichoanal syndrome. This condition involves several characteristic physical features, particularly affecting the eyes (oculo-), hair (tricho-), and anus (-anal). The mutations delete genetic material from the *FREM1* gene or result in a premature stop signal that leads to an abnormally short FREM1 protein, and are believed to result in a nonfunctional protein.

Absence of functional FREM1 protein interferes with its role in embryonic basement membrane development and may also affect the location, stability, or function of the FRAS1 and FREM2 proteins. The features of Manitoba oculotrichoanal syndrome may result from the failure of neighboring embryonic tissues to fuse properly due to impairment of the basement membranes' anchoring function.

other disorders

At least three *FREM1* gene mutations have been identified in people with a disorder called bifid nose, renal agenesis, and anorectal malformations syndrome, sometimes called BNAR. These mutations change single protein building blocks (amino acids) in the FREM1 protein or result in an abnormally shortened protein.

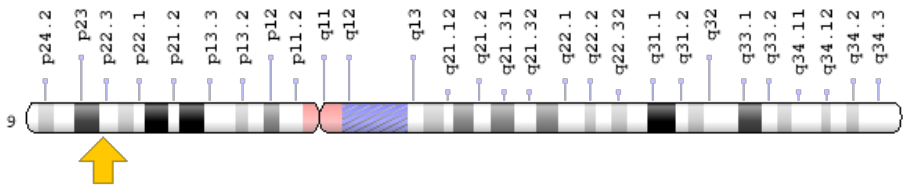
The mutations that cause BNAR likely disrupt the role of the FREM1 protein in the embryonic basement membranes of particular tissues, resulting in the cleft nasal cartilage (bifid nose), missing kidneys (renal agenesis), narrowed or misplaced anal opening (anorectal malformations), and other features characteristic of this disorder.

Researchers suggest that BNAR and Manitoba oculotrichoanal syndrome, with their overlapping features, may be considered part of a single disorder spectrum.

Chromosomal Location

Cytogenetic Location: 9p22.3, which is the short (p) arm of chromosome 9 at position 22.3

Molecular Location: base pairs 14,734,666 to 14,911,653 on chromosome 9 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- BNAR
- C9orf143
- C9orf145
- C9orf154
- extracellular matrix protein QBRICK
- FLJ25461
- FRAS1-related extracellular matrix protein 1
- FREM1_HUMAN
- TILRR

Additional Information & Resources

GeneReviews

- Manitoba Oculotrichoanal Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1728

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28FREM1%5BTIAB%5D%29+OR+%28%28BNAR%5BTIAB%5D%29+OR+%28TILRR%5BTIAB%5D%29+OR+%28C9orf14%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- BIFID NOSE WITH OR WITHOUT ANORECTAL AND RENAL ANOMALIES
  http://omim.org/entry/608980
- FRAS1-RELATED EXTRACELLULAR MATRIX PROTEIN 1
  http://omim.org/entry/608944

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=FREM1%5Bgene%5D
- HGNC Gene Family: C-type lectin domain containing
  http://www.genenames.org/cgi-bin/genefamilies/set/1298
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=23399
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/158326
- UniProt
  http://www.uniprot.org/uniprot/Q5H8C1

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